Case Report

Goldenhar Syndrome: A Case Report

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ABSTRACT

Goldenhar Syndrome, also known as Oculo-auriculo-vertebral Dysplasia, is one of the rare congenital developmental disorders that usually affects the anatomical structures derived from the first and second branchial arches. It is mostly sporadic in nature. Abnormality or defect in vascular supply of the embryo, as well as disruption in mesodermal migration, leads to the defective formation of the branchial arches and the vertebral system, which ultimately results in the development of Goldenhar syndrome. Clinical manifestations of Goldenhar Syndrome range from mild to severe forms. The characteristic clinical features are- maxillomandibular hypoplasia, epibulbar dermoid cyst, pretragal fistula, preauricular skin appendages, atresia of external, middle, and internal ear, and vertebral abnormalities. Here, we report a case of an 18-year-old male patient with typical orofacial manifestations of this syndrome. He had features like maxillomandibular hypoplasia, deviated nasal septum, preauricular tags, microtia, torticollis, cleft lip, and palate. The importance of the case report is to highlight the highly variable presentation of this congenital anomaly, facilitating early diagnosis and a multidisciplinary approach.

Key words: Oculo-Auriculo-Vertebral Spectrum, Congenital abnormalities, Craniofacial Abnormalities

Goldenhar Syndrome, also known as Oculo-auriculo-vertebral Dysplasia, is one of the rare congenital developmental disorders that usually affects the anatomical structures derived from the first and second branchial arches. In 1881, the characteristic asymmetrical malformations of this syndrome were first recorded by Von Arlt [1]. Later, in 1952, French ophthalmologist Dr. Maurice Goldenhar described the clinical features of this syndrome [2]. Later in 1963, Gorlin et al described an additional feature of vertebral anomalies in this syndrome and suggested the term 'Oculo-auriculo-vertebral dysplasia' [3]. In 1990, they combined several syndromes like Goldenhar Syndrome, Facioauriculovertebral syndrome, and hemifacial microsomia, with the first and second branchial arch anomalies, proposing the term Oculoauriculovertebral Spectrum [4].

The incidence rate of Goldenhar Syndrome is from 1:3500 to 1:5600 live births and 1:1000 in the case of children with congenital deafness. The male-to-female predilection ratio of this syndrome is 3:2 [5]. Most of the Goldenhar Syndrome cases are sporadic. However, multifactorial modes of inheritance have also been reported. Abnormality or defect in vascular supply of the embryo, disruption in mesodermal migration, can lead to the defective formation of the branchial arches and the vertebral system [6]. Chromosomal abnormalities like trisomy of 7, 9, or 22 chromosomes, translocations between chromosomes 5 and 8 have been reported in Goldenhar Syndrome [7]. Mounded et al reported a case where the mother had a history of vitamin A intoxication, leading to its teratogenic effect [8]. Other potential etiological factors of Goldenhar syndrome are the use of drugs such as thalidomide, retinoic acid, tamoxifen, and cocaine during the gestational period, infection of rubella, influenza during pregnancy, and gestational diabetes [3].

Clinical manifestations of Goldenhar Syndrome range from mild to severe forms. The characteristic clinical features are- maxillomandibular hypoplasia, zygomatic bone hypoplasia, epibulbar dermoid, ptosis, ocular hypertelorism, anophthalmos, pretragal fistula, preauricular skin appendages, microtia, atresia of external, middle, internal ear, coloboma of upper eyelid, antimongoloid palpebral fissures, cleft lip and palate, bifid tongue, facial nerve involvement, plaque induced gingival hypertrophy due to poor oral hygiene [9]. In the present case, an 18-year-old male patient having dental pain reported to the outpatient department and had the clinical features like maxillomandibular hypoplasia, microtia, lagophthalmos, cleft lip and palate, deviated nasal septum, etc., diagnosed as Goldenhar Syndrome.

CASE REPORT

An 18-year-old male patient reported to the outpatient

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department of Oral and Maxillofacial Pathology of a tertiary health care center with the chief complaint of pain in the lower left back tooth region for the last month.

After recording proper history, an extraoral clinical examination was done, revealing asymmetry of face along with hypoplasia of the maxillomandibular region, depressed left zygomatic arch, unilateral macrostomia, left ear microtia and rudimentary pinna (Figure 1a), lagophthalmos in left eye (Figure 1b), deviated nasal septum, prominent antegonial notch, prominent chin bone, deviated angle of mouth, antimongoloid slanting of left eye, preauricular tags. He had partial hearing impairment in the left ear; the right ear was not affected.

Figure 1. a. Asymmetry of the face along with hypoplasia of the maxillomandibular region with left ear microtia and rudimentary pinna.

b. Lagophthalmos in the left eye

There was no family history of these features. The patient had three siblings, but none of them had any abnormalities. At the time of his birth, the paternal and maternal ages were 34 years and 27 years, respectively. The patient had no systemic abnormality. Torticollis and cleft lip surgery were done at the age of 3 months.

During mouth opening, the mandible deviated towards the left. Slurring of speech was noted. Left-sided facial nerve palsy (Figure 1b) was another prominent feature. Torticollis was present at birth, which was resolved after surgery. Intraoral examination revealed a high arched palate, crowding of teeth, multiple carious teeth, retained deciduous left mandibular canine, posterior open bite, and posterior crossbite (Figure 2). The patient had pain with respect to the grossly carious mandibular left permanent molar, which was non-restorable, hence was indicated for extraction and replacement.

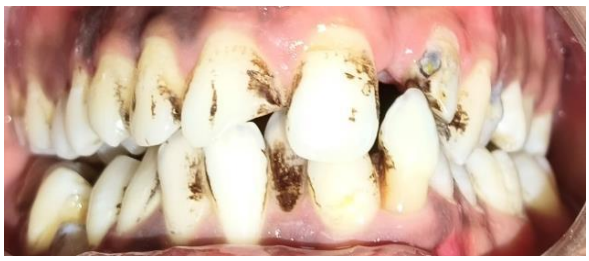

Figure 2. Crowding of teeth, retained deciduous left mandibular canine, posterior open bite and posterior crossbite

A history of cleft lip and cleft palate was present, and the cleft lip had been resolved by surgery at the age of 3 months. A deviated nasal septum was present and was corrected by conchal graft. The patient had hearing impairment on the affected side and delayed motor development. However, the patient had no behavioral abnormalities.

Orthopantomogram (OPG) showed maxillomandibular hypoplasia on the left side, prominent antegonial notch, crowding, and multiple carious teeth. To further analyze the findings, a High-Resolution Computed Tomography (HRCT) scan of the temporal bone was done, which showed left external auditory canal atresia, a rudimentary middle ear cavity, an absent ossicular chain, and an altered facial nerve canal. Three-dimensional Non-Contrast Computed Tomography (NCCT) of the face showed a deviated nasal septum (Figure 3a), a 7x9 mm defect in the alveolar process and anterior hard palate on the left side (Figure 3b).

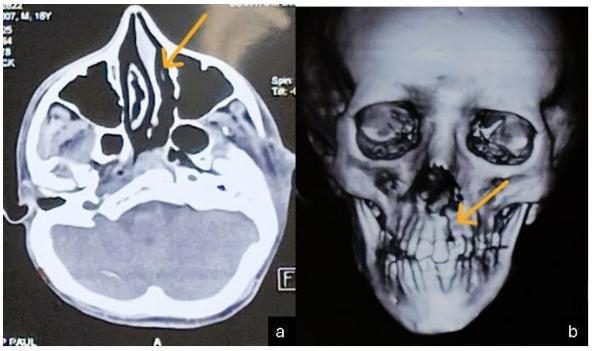

Figure 3. NCCT of the face showing.

a. Deviated nasal septum (yellow arrow)
b. Defect in the alveolar process and anterior hard palate on the left side (yellow arrow)

The common differential diagnoses include Treacher-Collins Syndrome, Townes-Brocks Syndrome, Wolf-Hirschhorn Syndrome, Delleman Syndrome, Nager Syndrome, and VACTERL Syndrome. Based on the patient's history, clinical examination, and investigations, a diagnosis of Goldenhar syndrome was made. The Extraction of the grossly carious mandibular left permanent molar was done to relieve the patient's complaints of pain.

DISCUSSION

In the present case, an 18-year-old patient was diagnosed with Goldenhar Syndrome, and the associated clinical features were unilateral facial asymmetry, maxillomandibular hypoplasia, unilateral macrostomia, microtia, rudimentary pina, deviated nasal septum, lagophthalmos, cleft lip and palate, torticollis, etc., with an unknown etiology. In around 85% cases, the abnormalities are limited to the unilateral aspect, whereas 10-33% cases have bilateral manifestation [5]. The right side is usually more frequently affected, although in this present case, the left side is involved. Facial nerve involvement is commonly reported, which was also present in the present

case [5]. 40-60% of patients with Goldenhar Syndrome have vertebral anomalies such as the presence of wedge-shaped vertebrae, spina bifida, scoliosis, synostosis, etc., as reported by Gorlin et al [10]. Cardiovascular alterations like Tetralogy of Fallot and Ventricular Septal Defect have been reported in about 5-58% of cases. Other systemic abnormalities may include mental retardation, tracheoesophageal fistula, and respiratory illness [6]; torticollis [11], ectopic kidneys, CNS occipital encephalocele, developmental delay, inguinal hernia, umbilical hernia, etc. [12].

In the current case, the patient had no systemic complication except torticollis, which was corrected by surgery at the age of 3 months. Das A et al. reported a case where a rare feature was associated with this syndrome; right-sided hypoplastic thumb with flattened thenar muscle, resembling "floating" thumb [13]. In the present case, no such limb anomaly was present. Jena KA reported about an attack of epilepsy because of localized calcification in the occipital and parietal region of the brain and hypertrophy of the left hemisphere, unilateral complex odontoma in both jaws [14]. Martinelli et al reported ipsilateral cerebellar hypoplasia in a fetus with oculoauriculovertebral spectrum. In the case reported, neurological abnormalities were absent [15]. The diagnosis of Goldenhar Syndrome is mostly clinical; however, investigations like imaging, Computed Tomography (CT) scan, Magnetic Resonance Imaging (MRI), Electroencephalogram (EEG), ultrasound (USG), and echocardiogram (ECHO) may be needed for proper assessment and grading of the severity of the signs and symptoms [16].

Treatment of patients with Goldenhar syndrome requires multidisciplinary team management. In uncomplicated cases without systemic involvement, the prognosis is good. Cosmetic surgery can be performed to correct structural anomalies of the ears, eyes, and nose. A multidisciplinary approach includes surgical repair of the defect of cleft lip and palate, excision of dermoids, reconstruction surgery with the help of rib bone graft for correction of hypoplastic mandible, distraction osteogenesis for maxillary bone augmentation, and postoperative orthodontic management. Constant follow-up and reassessment of results are essential [17].

CONCLUSION

Dental practitioners should have extensive knowledge about the clinical manifestations and systemic findings associated with Goldenhar syndrome. The significance of this case presentation is that the variable clinical features associated with this syndrome will facilitate early diagnosis, multidisciplinary management, and guidance to the patient to achieve a satisfactory functional and aesthetic outcome.

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